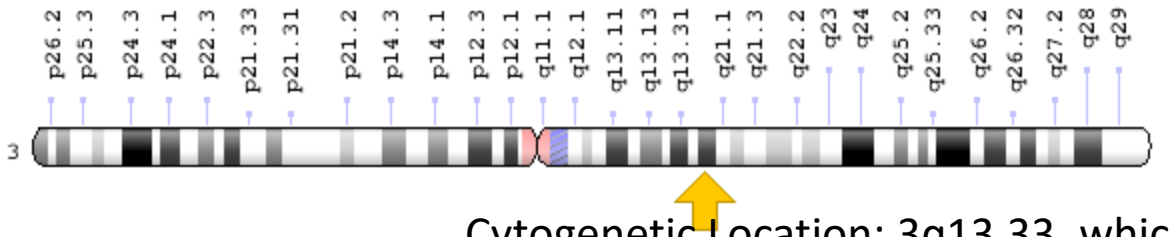
alkaptonuria

- HGD
- homogentisate 1,2-dioxygenase

- The *HGD* gene provides instructions for making an enzyme called homogentisate oxidase, which is active chiefly in the liver and kidneys. This enzyme participates in a step-wise process that breaks down two protein building blocks (amino acids), phenylalanine and tyrosine, when they are no longer needed or are present in excess. These two amino acids also play a role in making certain hormones, pigments, and brain chemicals called neurotransmitters.

- Homogentisate oxidase is responsible for a specific step in the breakdown of phenylalanine and tyrosine. Previous steps convert the two amino acids into a molecule called homogentisic acid. Homogentisate oxidase adds two oxygen atoms to homogentisic acid, converting it to another molecule called maleylacetoacetate. Other enzymes break down maleylacetoacetate into smaller molecules that are later used for energy or to make other products that can be used by the body.



Cytogenetic Location: 3q13.33, which is the long (q) arm of chromosome 3 at position 13.33

Molecular Location: base pairs 120,628,168 to 120,682,571 on chromosome 3 (Homo sapiens Annotation Release 108, GRCh38.p7)

- Mutations in the HGD gene can cause the rare disorder alkaptonuria. The gene usually produces an enzyme that breaks down a substance called homogentisic acid. When this enzyme doesn't function properly, the homogentisic acid builds up and causes symptoms of alkaptonuria that include arthritis, black urine and reddish earwax

other names people used for alkaptonuria

- AKU
- alcaptonuria
- homogentisic acid oxidase deficiency
- homogentisic acidura

- Alkaptonuria is a rare condition in which a person's urine turns a dark brownish-black color when exposed to air. Alkaptonuria is part of a group of conditions known as an inborn error of metabolism
- Alkaptonuria is caused by mutations in the HGD gene. It is inherited in an autosomal recessive fashion.

- The gene defect makes the body unable to properly break down certain amino acids (tyrosine and phenylalanine). As a result, a substance called homogentisic acid builds up in the skin and other body tissues. The acid leaves the body through the urine. The urine turns brownish-black when it mixes with air.

- Alkaptonuria is inherited, which means it is passed down through families. If both parents carry a nonworking copy of the gene related to this condition, each of their children has a 25% (1 in 4) chance of developing the disease.

- Urine in an infant's diaper may darken and can turn almost black after several hours. However, many people with this condition may not know they have it until mid-adulthood (around age 40), when joint and other problems occur.

- Symptoms may include:
- Arthritis (especially of the spine) that gets worse over time
- Darkening of the ear
- Dark spots on the white of the eye and cornea
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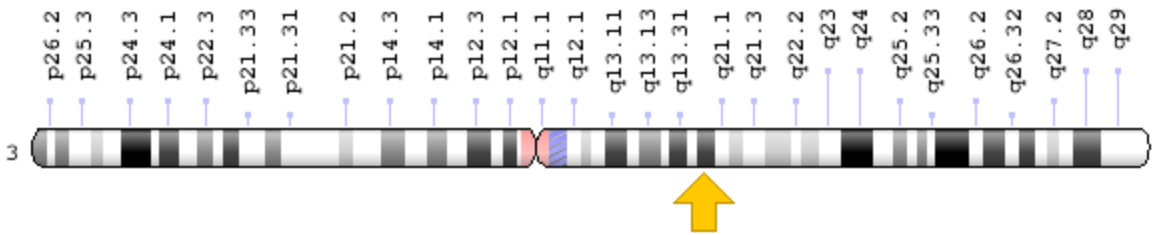

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- This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

- Mutations in the HGD gene cause alkaptonuria. The HGD gene provides instructions for making an enzyme called homogentisate oxidase. This enzyme helps break down the amino acids phenylalanine and tyrosine, which are important building blocks of proteins. Mutations in the HGD gene impair the enzyme's role in this process. As a result, a substance called homogentisic acid, which is produced as phenylalanine and tyrosine are broken down, accumulates in the body.

- Excess homogentisic acid and related compounds are deposited in connective tissues, which causes cartilage and skin to darken. Over time, a buildup of this substance in the joints leads to arthritis. Homogentisic acid is also excreted in urine, making the urine turn dark when exposed to air.

- Alkaptonuria is an inherited condition that causes urine to turn black when exposed to air. Ochronosis, a buildup of dark pigment in connective tissues such as cartilage and skin, is also characteristic of the disorder. This blue-black pigmentation usually appears after age 30. People with alkaptonuria typically develop arthritis, particularly in the spine and large joints, beginning in early adulthood. Other features of this condition can include heart problems, kidney stones, and prostate stones.

- This condition is rare, affecting 1 in 250,000 to 1 million people worldwide. Alkaptonuria is more common in certain areas of Slovakia (where it has an incidence of about 1 in 19,000 people) and in the Dominican Republic.